



Wagner syndrome

Wagner syndrome is a hereditary disorder that causes progressive vision loss. The eye problems that lead to vision loss typically begin in childhood, although the vision impairment might not be immediately apparent.

In people with Wagner syndrome, the light-sensitive tissue that lines the back of the eye (the retina) becomes thin and may separate from the back of the eye (retinal detachment). The blood vessels within the retina (known as the choroid) may also be abnormal. The retina and the choroid progressively break down (degenerate). Some people with Wagner syndrome have blurred vision because of ectopic fovea, an abnormality in which the part of the retina responsible for sharp central vision is out of place. Additionally, the thick, clear gel that fills the eyeball (the vitreous) becomes watery and thin. People with Wagner syndrome develop a clouding of the lens of the eye (cataract). Affected individuals may also experience nearsightedness (myopia), progressive night blindness, or a narrowing of their field of vision.

Vision impairment in people with Wagner syndrome can vary from near normal vision to complete loss of vision in both eyes.

Frequency

Wagner syndrome is a rare disorder, although its exact prevalence is unknown. Approximately 300 affected individuals have been described worldwide; about half of these individuals are from the Netherlands.

Genetic Changes

Mutations in the *VCAN* gene cause Wagner syndrome. The *VCAN* gene provides instructions for making a protein called versican. Versican is found in the extracellular matrix, which is the intricate lattice of proteins and other molecules that forms in the spaces between cells. Versican interacts with many of these proteins and molecules to facilitate the assembly of the extracellular matrix and ensure its stability. Within the eye, versican interacts with other proteins to maintain the structure and gel-like consistency of the vitreous.

VCAN gene mutations that cause Wagner syndrome lead to insufficient levels of versican in the vitreous. Without enough versican to interact with the many proteins of the vitreous, the structure becomes unstable. This lack of stability in the vitreous affects other areas of the eye and contributes to the vision problems that occur in people with Wagner syndrome. It is unknown why *VCAN* gene mutations seem solely to affect vision.

Inheritance Pattern

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Other Names for This Condition

- hyaloideoretinal degeneration of Wagner
- VCAN-related vitreoretinopathy
- Wagner disease
- Wagner vitreoretinal degeneration
- Wagner vitreoretinopathy

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Wagner syndrome
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0339540/>

Other Diagnosis and Management Resources

- GeneReview: VCAN-Related Vitreoretinopathy
<https://www.ncbi.nlm.nih.gov/books/NBK3821>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Health Topic: Cataract
<https://medlineplus.gov/cataract.html>
- Health Topic: Retinal Disorders
<https://medlineplus.gov/retinaldisorders.html>
- Health Topic: Vision Impairment and Blindness
<https://medlineplus.gov/visionimpairmentandblindness.html>

Genetic and Rare Diseases Information Center

- Wagner syndrome
<https://rarediseases.info.nih.gov/diseases/7871/wagner-syndrome>

Additional NIH Resources

- National Eye Institute: Diagram of the Eye
<https://nei.nih.gov/health/eyediagram/>
- National Eye Institute: Low Vision
<https://nei.nih.gov/health/lowvision/>

Educational Resources

- Centers for Disease Control and Prevention: Vision Impairment
https://www.cdc.gov/ncbddd/actearly/pdf/parents_pdfs/VisionLossFactSheet.pdf
- Cleveland Clinic: Cataracts and Cataract Surgery
<http://my.clevelandclinic.org/health/articles/cataracts>
- Cleveland Clinic: Coping with Vision Loss
<http://my.clevelandclinic.org/health/articles/coping-vision-loss>
- Disease InfoSearch: Wagner syndrome
<http://www.diseaseinfosearch.org/Wagner+syndrome/7430>
- MalaCards: wagner syndrome
http://www.malacards.org/card/wagner_syndrome
- Merck Manual Consumer Version: Overview of Retinal Disorders
<http://www.merckmanuals.com/home/eye-disorders/retinal-disorders/overview-of-retinal-disorders>
- Orphanet: Wagner disease
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=898

Patient Support and Advocacy Resources

- American Foundation for the Blind
<http://www.afb.org/>
- Prevent Blindness America
<http://www.preventblindness.org/>
- Retina International
<http://www.retina-international.org/>
- The Foundation Fighting Blindness
<http://fffb.ca/>
- University of Kansas Medical Center Resource List: Blind/Visual Impairment
<http://www.kumc.edu/gec/support/visual.html>

GeneReviews

- VCAN-Related Vitreoretinopathy
<https://www.ncbi.nlm.nih.gov/books/NBK3821>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28wagner+syndrome%5BTIAB%5D%29+OR+%28wagner+disease%5BTIAB%5D%29+NOT+%28Stickler%5BTI%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- WAGNER VITREORETINOPATHY
<http://omim.org/entry/143200>

Sources for This Summary

- GeneReview: VCAN-Related Vitreoretinopathy
<https://www.ncbi.nlm.nih.gov/books/NBK3821>
- Kloeckener-Gruissem B, Bartholdi D, Abdou MT, Zimmermann DR, Berger W. Identification of the genetic defect in the original Wagner syndrome family. *Mol Vis*. 2006 Apr 17;12:350-5.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/16636652>
- Meredith SP, Richards AJ, Flanagan DW, Scott JD, Poulson AV, Snead MP. Clinical characterisation and molecular analysis of Wagner syndrome. *Br J Ophthalmol*. 2007 May;91(5): 655-9. Epub 2006 Oct 11.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/17035272>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1954774/>

- Miyamoto T, Inoue H, Sakamoto Y, Kudo E, Naito T, Mikawa T, Mikawa Y, Isashiki Y, Osabe D, Shinohara S, Shiota H, Itakura M. Identification of a novel splice site mutation of the CSPG2 gene in a Japanese family with Wagner syndrome. Invest Ophthalmol Vis Sci. 2005 Aug;46(8):2726-35.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/16043844>
- Mukhopadhyay A, Nikopoulos K, Maugeri A, de Brouwer AP, van Nouhuys CE, Boon CJ, Perveen R, Zegers HA, Wittebol-Post D, van den Biesen PR, van der Velde-Visser SD, Brunner HG, Black GC, Hoyng CB, Cremers FP. Erosive vitreoretinopathy and wagner disease are caused by intronic mutations in CSPG2/Versican that result in an imbalance of splice variants. Invest Ophthalmol Vis Sci. 2006 Aug;47(8):3565-72.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/16877430>
- Ronan SM, Tran-Viet KN, Burner EL, Metlapally R, Toth CA, Young TL. Mutational hot spot potential of a novel base pair mutation of the CSPG2 gene in a family with Wagner syndrome. Arch Ophthalmol. 2009 Nov;127(11):1511-9. doi: 10.1001/archophthalmol.2009.273.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19901218>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3514888/>
- OMIM: WAGNER VITREORETINOPATHY
<http://omim.org/entry/143200>

Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/condition/wagner-syndrome>

Reviewed: July 2014

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications

U.S. National Library of Medicine

National Institutes of Health

Department of Health & Human Services